About Yourgene Biosciences
Yourgene Biosciences is part of the Premaitha Health plc group of companies, a leading international molecular diagnostics group. Our primary focus is using the latest advances in DNA analysis technology to develop safer, faster and more accurate non-invasive prenatal screening tests for pregnant women.

Sage™ prenatal screen reduces the number of unnecessary invasive tests such as amniocentesis, which carry a small risk of miscarriage. However, it is a screening test and all high-risk results should be confirmed by a follow-up invasive procedure.

The Sage™ prenatal screen is safe, fast and accurate: giving you and your family peace of mind.

NIPT considerations:

• **Fetal Fraction**
  Some pregnant women have a lower proportion of placental DNA in their blood; this is called a “low fetal fraction”. This can be due to several different reasons; for example, women with a high maternal weight may have increased blood volume which could result in a dilution of the cell free placental DNA in the mother’s plasma. Some NIPTs will not be sensitive enough to generate an accurate result on samples with decreased fetal fraction. The Sage™ prenatal screen has an adaptive software that is able to produce a valid and accurate result in samples that have as little as 3.5% fetal fraction.

• **Re-draw rate**
  NIPTs may not always be able to generate a test result from the blood sample and a re-draw will be required. The Sage™ prenatal screen has one of the lowest re-draw rates (less than 1 in 100 samples). If this occurs a second blood sample may be required and will be tested at no additional cost.

• **Turnaround time to results**
  Waiting for screening test results is an anxious and stressful time for you and your family. With the Sage™ prenatal screen we are committed to ensuring your results are received by your healthcare provider as fast as possible. The majority of Sage™ prenatal screens are analysed in as little as 3-5 days from sample receipt.

• **IVF, surrogate and donor pregnancies**
  The Sage™ prenatal screens is suitable for IVF, surrogate and egg donor pregnancies, unlike some NIPTs.

For the latest news and updates about the Sage prenatal screen please follow us on:  

/SAGEprenatalscreen  @SAGEprenatal

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YMK001 Rev A
Autosomal aneuploidies

Trisomies occur when three, instead of the usual two, copies of a chromosome are present in each cell. When the number of chromosomes differs from the usual two this is referred to as an aneuploidy. The Sage™ prenatal screen test estimates the risk of a fetus having Down’s syndrome (Trisomy 21), Edwards’ syndrome (Trisomy 18) and Patau’s syndrome (Trisomy 13). Each person also has a set of sex chromosomes in each cell. Women have two ‘X’ chromosomes (XX) while men have one ‘X’ and one ‘Y’ chromosome (XY). The impact of sex chromosome aneuploidy is generally much milder than aneuploidy of chromosomes 13, 18, and 21. The following aneuploidies can be screened for:

- Monosomy X – Turner syndrome
- XXX – Triple X syndrome
- XXY – Kleinfelter syndrome
- XYY – Jacob’s syndrome

Sex chromosome aneuploidies

In addition to the autosomal trisomies, the Sage™ prenatal test also may also screen for sex chromosomal aneuploidies. Women have two ‘X’ chromosomes (XX) while men have one ‘X’ and one ‘Y’ chromosome (XY). The impact of sex chromosome aneuploidy is generally much milder than aneuploidy of chromosomes 13, 18, and 21. The following aneuploidies can be screened for:

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Microdeletions

Upon request, testing for microdeletion syndromes is available. A microdeletion syndrome is caused by the absence of a small portion of genetic material in the chromosome. They vary greatly in severity, with the symptoms of microdeletions ranging from minimal developmental delays to severe anomalies e.g. cardiac defects, neurological malformations, etc.

What are the advantages of the Sage™ prenatal screen?

**Safe:** Non-invasive with no risk of miscarriage

**Fast:** The Sage™ prenatal screen is one of the fastest NIPT available with results provided within just 3-5 working days, from sample receipt.

**Accurate:** Greater than 99% detection of autosomal trisomy conditions with a low redraw rate of 1 in 100.

**Simple:** Uses just one simple maternal blood sample of 10ml

**In depth:** In addition to the common trisomies 21, 18 and 13, a whole chromosome analysis is available upon request, along with sex chromosome aneuploidies and microdeletions.

What does Sage™ screen for?

**Autosomal aneuploidies**

The Sage™ prenatal screen is a non-invasive prenatal test (NIPT) for pregnant women which estimates the risk of a fetus having Down’s syndrome or other serious genetic diseases. The Sage™ prenatal screen is an advanced screening test that is carried out on a small maternal blood sample. Pregnant women can expect test results from their healthcare providers within 3-5 working days from sample receipt.

**Sex chromosome aneuploidies**

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How does it work?

During pregnancy, the placenta leaks fetal cell-free DNA which circulates in the maternal bloodstream. As a result, a maternal blood sample contains a mixture of fetal and maternal circulating DNA. The Sage™ prenatal screen directly measures the amount of this cell-free DNA and can detect small changes in the DNA ratio between the maternal and cell-free DNA to estimate the risk of a fetal chromosomal aneuploidy or a microdeletion being present.

Who can have the Sage™ prenatal screen?

- Suitable for women who are at least 10 weeks pregnant
- Suitable for all singleton and twin pregnancies
- Suitable for all IVF and surrogate pregnancies
- Unsuitable for women with cancer, a trisomy, have undergone a blood transfusion within the last 12 months or have ever undergone stem cell therapy, immunotherapy or received an organ transplant

How are the Sage™ prenatal screen results reported?

- **Low Risk:** means that it is very unlikely your pregnancy is affected by the trisomy or aneuploidy.
- **High risk:** means that your pregnancy is at increased risk for trisomy or aneuploidy and the result should be confirmed by a follow up invasive such as amniocentesis.
- **No result:** Very occasionally there is insufficient placental DNA in the sample to obtain a result. Women may be asked back by your healthcare provider for a further blood sample.

**Sage™ prenatal screening pathway**

- **Screening method 1:** Traditional first trimester screening Accuracy 85%
- **Screening method 2:** Non-invasive prenatal screening Accuracy >99%
- **Screening method 3:** Invasive diagnostics Accuracy - 100%

**First phase screening**

- NIP: Sage™ prenatal screen (from 10 weeks gestation)

**Second phase screening**

- NIPT: Sage™ prenatal screen (from 10 weeks gestation)

**Amniocentesis from 16 weeks gestation**

**Chorionic villus sampling (CVS) from 14 weeks gestation**

**Results sent to healthcare professional**

• Low risk: No further testing required.

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